PARK7 gene

Parkinsonism associated deglycase

Normal Function

The *PARK7* gene provides instructions for making the DJ-1 protein. This protein is found in many tissues and organs, including the brain. Studies indicate that the DJ-1 protein has several functions, although none are fully understood. One of the protein's functions may be to help protect cells, particularly brain cells, from oxidative stress. Oxidative stress occurs when unstable molecules called free radicals accumulate to levels that can damage or kill cells. Additionally, the DJ-1 protein may serve as a chaperone molecule that helps fold newly produced proteins into the proper 3-dimensional shape and helps refold damaged proteins. Like other chaperone molecules, the DJ-1 protein may assist in delivering selected proteins to proteasomes, which are structures within cells that break down unneeded molecules. Researchers suggest that the DJ-1 protein may also play a role in activities that produce and process RNA, a chemical cousin of DNA.

Health Conditions Related to Genetic Changes

Parkinson disease

Researchers have identified more than 25 *PARK7* gene mutations that can cause Parkinson disease, a condition characterized by progressive problems with movement and balance. These mutations are associated with the early-onset form of the disorder, which begins before age 50. Some *PARK7* gene mutations lead to an abnormally small DJ-1 protein or change the building blocks (amino acids) used to make the protein. The altered protein is unstable and does not function properly, if at all. Other mutations delete a large portion of the *PARK7* gene, preventing the production of any functional DJ-1 protein.

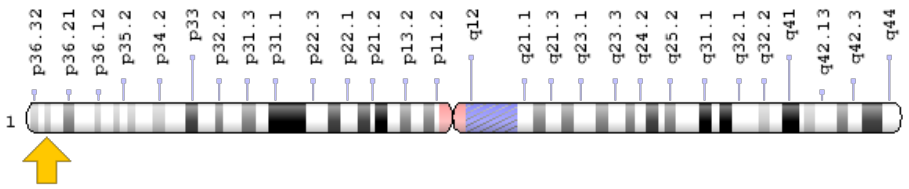
It is unclear how loss of functional DJ-1 protein leads to Parkinson disease. Some studies suggest that *PARK7* gene mutations disrupt the protein's chaperone function, which leads to a toxic buildup of misfolded or damaged proteins and eventually to cell death. Another possibility is that *PARK7* gene mutations impair the protein's ability to protect cells from destructive oxidative stress. Nerve cells that make the chemical messenger dopamine are particularly vulnerable to oxidative stress. With diminished protection, free radicals may cause enough damage to kill these nerve cells. Progressive loss of dopamine-producing nerve cells is a characteristic feature of Parkinson disease. The death of these cells weakens communication between

the brain and muscles, and ultimately the brain becomes unable to control muscle movement.

Chromosomal Location

Cytogenetic Location: 1p36.23, which is the short (p) arm of chromosome 1 at position 36.23

Molecular Location: base pairs 7,961,654 to 7,985,282 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DJ-1
- DJ1
- PARK7_HUMAN
- Parkinson disease (autosomal recessive, early onset) 7
- parkinson protein 7

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: The PARK7 Locus and the DJ-1 Gene https://www.ncbi.nlm.nih.gov/books/NBK6225/#A28771
- Parkinson's Disease: National Clinical Guideline for Diagnosis and Management in Primary and Secondary Care (2006): Neuroprotection https://www.ncbi.nlm.nih.gov/books/NBK48504/#ch6.s1

GeneReviews

- Parkinson Disease Overview https://www.ncbi.nlm.nih.gov/books/NBK1223

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28PARK7%5BTIAB%5D%29+OR+%28DJ1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

OMIM

- ONCOGENE DJ1
  http://omim.org/entry/602533

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/PARK7ID41639ch1p36.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PARK7%5Bgene%5D
- HGNC Gene Family: Glutamine amidotransferase like class 1 domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/1408
- HGNC Gene Family: Parkinson disease associated genes
  http://www.genenames.org/cgi-bin/genefamilies/set/672
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16369
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/11315
- PDGene
  http://www.pdgene.org/view?gene=PARK7
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